

Illinois Department of Public Health  
Newborn Screening Laboratory Subcommittee  
Illinois Department of Public Health, Division of Laboratories  
2121 W. Taylor St., Chicago, Illinois  
Meeting and Conference Call Minutes: September 18, 2013

Subcommittee Members Attending:

George Hoganson-University of Illinois at Chicago – Chair  
Gopal Srinivasan- Mt. Sinai Hospital  
Denise Lonigro-Advocate Christ Hospital  
Kristin Clemenz-Lurie Children’s Memorial  
W. Patrick Zeller- Endocrinologist- Private Practice

IDPH Staff:

George Dizikes, Arthur Kohrman, Khaja Basheeruddin, Rong Shao, Raj Singh, Jennifer Crew, Joel Price, Claudia Nash, Shannon Harrison, Jean Becker

The meeting was called to order at 9:05 AM, followed by introductions. The minutes were approved for the meeting held February 27, 2013. A quorum was not present for the subsequent meeting scheduled for June 5, 2013.

**Old Business**

There were no items for discussion.

**New Business**

**Laboratory Report**

Staffing and Laboratory Resources:

Dr. Dizikes indicated that space on the second floor of the Chicago lab was renovated and all newborn screening operations except the original MS/MS lab has been consolidated and moved to this floor, which will greatly improve the efficiency of the operation. Laboratory staffing is stable and there is sufficient coverage, although some staff are considering retirement. Cross training is occurring within the unit. Staff in the molecular unit who will be performing SCID testing are learning additional responsibilities, such as specimen punching. The overall number of galactosemia positives has drastically decreased this summer compared with the prior two years. It is not certain what factors contributed to this change in samples with reduced GALT enzyme activity.

Data System/Reports:

Dr. Dizikes indicated that progress continues with the implementation of electronic transfer of data between Northwestern Memorial Hospital and the IDPH Newborn Screening Laboratory through an HL7 interface. Since the greatest number of births in the state occur at this facility, Northwestern was selected as the first hospital to implement this method of electronic data exchange, which should be piloted in the near future. It was stated that over half of the Illinois birth hospitals are using Epic for electronic medical records but it was stated that for each hospital, further modification will likely be required to interface with the IDPH newborn screening module.

Dr. Dizikes also stated that the contract has been finalized with Perkin Elmer for the current fiscal year, and that there has been some progress made towards “eReports” with the Perkin Elmer data system, where health care providers will be able to access the IDPH newborn screening data system to obtain test results for their patients.

Lysosomal Storage Disease (LSD) and Severe Combined Immune Deficiency (SCID) Screening:

Dr. Dizikes reported that three tandem mass spectrometers have been installed which will be used for LSD testing. Validation is continuing on the new testing methods and specimen exchange with Perkin Elmer for LSD and SCID will occur in late winter or early spring with the target date for statewide implementation of both tests still anticipated as July 1, 2014. Current newborn screening educational materials have been revised to include information on LSDs and SCID.

Non-Derivatized Amino acid and Carnitine Testing

Proficiency and quality control testing has been performed already at the IDPH lab with the non-derivatized method for amino acid and acylcarnitine analytes. The lab is awaiting the delivery of the new tandem mass spectrometers which will be obtained from Perkin Elmer. It is anticipated that use of this new method should occur after SCID and LSD testing have been implemented.

The new non-derivatized method will allow succinylacetone testing on all samples, which will help identify possible cases of tyrosinemia Type I. Dr. Hoganson noted that there has been an increase in the number of newborns in the neonatal intensive care unit with high tyrosine. This may be due to changes in NICU feeding practices, where these babies are getting more tyrosine in their diet. Currently, changes to the laboratory report to more clearly indicate succinylacetone values are being considered and the proposed language will be distributed to the metabolic specialists and to members of this committee for their review.

Endocrine Testing

The IDPH Newborn Screening Laboratory expects to take have a new GSP instrument delivered from Perkin Elmer this week, which it will evaluate. The GSP will eventually replace the AutoDELFIA for endocrine testing and will also eventually perform testing for galactosemia (total galactose and GALT) and biotinidase deficiency. This switchover will likely occur after the implementation of LSD, SCID and non-derivatized MS/MS testing are in place.

Hemoglobinopathy Testing

There are no issues or concerns currently with newborn screening for sickle cell and related conditions.

**Follow-up Program Report**

Staffing

Three newborn screening staff have accepted positions elsewhere at IDPH . One newborn screening follow-up position will be replaced October 1, and the second similar position has not yet been posted. A third newborn screening follow-up person should be hired when SCID testing occurs. The nurse supervisor positions that was vacated in August has not yet been approved for posting. Shannon Harrison, a nurse in the program has been temporarily assigned to also assume these duties in the interim along with her primary responsibilities of being the education coordinator.

Critical Congenital Heart Disease (CCHD):

Governor Quinn signed House Bill 2661 on August 16, 2013, which amended the Newborn Metabolic Screening Act to immediately require all birth hospitals to screen all newborns for critical congenital heart disease (CCHD), using pulse oximetry. IDPH has distributed to hospitals a recommended screening protocol that was developed by the IDPH CCHD Work Group that has met for the past year, and will work with the Illinois Chapter of the American Heart Association to develop and distribute additional patient educational resources. Newborn screening Administrative Rules will need to be amended to describe the specific requirements of screening implementation.

Other Issues

Further discussion of qualifications required for specialists as described in the Newborn Metabolic Screening and Treatment Code will be deferred until the October 31 meeting of the full Genetic and Metabolic Diseases Advisory Committee.

Dr. Srinivasan from Mt .Sinai brought up the issues of hospital newborn screening contact staff and it was clarified by the Follow-Up Program that the only reports requiring immediate follow up by the contact person are those that are highly elevated. Follow-Up staff will indicate which cases are high risk when they are reported. The possibility of requiring each hospital to designate a newborn screening contact person was also discussed. Currently about 20 of the larger birthing hospitals have a staff person to whom all abnormal newborn screening tests are reported, who then notify the primary care provider. While it is helpful to have a designated staff person at each hospital, if they are only assigned to this duty on a part time basis, it becomes problematic when IDPH need to report abnormal results and the contact person is not available. IDPH staff also reported that tracking submission of samples and receipt of final test results is a laboratory function at many hospitals, and that the newborn screening contact person at most hospitals is a nurse in labor and delivery.

The meeting was adjourned at 9:45 am.